

CLAIMS

1. A method for determining whether an individual has an enhanced, diminished, or average probability of exhibiting one or more phenotypic attributes, comprising:

5 evaluating genomic markers from an individual for zygosity at each member of a preselected set of markers;

10 comparing the markers to a multivariate scoring matrix to obtain a marker score, wherein the multivariate scoring matrix correlates patterns of marker zygosity with probabilities of exhibiting phenotypic attributes; and

15 determining whether the marker score indicates an enhanced, diminished, or average probability of exhibiting one or more phenotypic attributes.

2. The method according to claim 1, wherein the preselected set of markers comprises a plurality of exon/intron junction sequences.

20 3. The method according to claim 2, wherein at least about 20% of the markers in the preselected set are exon/intron junction sequences.

4. The method according to claim 1, wherein the preselected set of markers comprises a plurality of promoter sequences.

25 5. The method according to claim 4, wherein at least about 20% of the markers in the preselected set are promoter sequences.

30 6. The method according to claim 1, wherein one or more of the markers within the preselected set of markers is selected by prioritizing with respect to one or more criteria selected from the group consisting of nucleotide sequence homology, synteny with

respect to other marker sequences, ontological relevance, genomic relevance, quality of supporting research, and degree of phenotypic significance.

7. The method according to claim 1, wherein the preselected set of markers comprises markers that map to at least about 1,000 discrete loci.

8. The method according to claim 1, wherein the scoring matrix prioritizes markers with respect to one or more criteria selected from the group consisting of homology to another marker sequence of interest, synteny with respect to other marker sequences, ontological relevance, genomic relevance, and quality of supporting research.

9. A method for providing relevant genetic information to an individual, comprising:

identifying genotypic characteristics of the individual that correlate with a relative probability of exhibiting one or more phenotypic characteristics;

determining for each of the one or more phenotypic characteristics whether the individual has an enhanced, diminished, or average probability of exhibiting the characteristic by:

(i) evaluating genomic markers from an individual for zygosity at each member of a preselected set of markers;

(ii) comparing the markers to a multivariate scoring matrix to obtain a marker score, wherein the multivariate scoring matrix correlates patterns of marker zygosity with probabilities of exhibiting phenotypic attributes; and

(iii) determining whether the marker score indicates an enhanced, diminished, or average probability of exhibiting the one or more phenotypic attributes;

then applying one or more selection criteria for each of the one or more phenotypic characteristics to the resulting determinations of enhanced, diminished, or average probability, wherein each selection criterion imposes total, partial, or no limitation on the information communicated to the individual;

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identifying information that is relevant to the individual's probabilities of exhibiting the one or more phenotypic characteristics and consistent with the limitations imposed by the selection criteria; and

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communicating the information to the individual.

10. The method according to claim 9, further comprising:

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applying the same or different selection criteria one or more additional times to the determined probabilities of exhibiting each of the phenotypic characteristics;

identifying information that is relevant to the individual's probabilities of exhibiting the one or more phenotypic characteristics and consistent with the limitations imposed by the selection criteria; and

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communicating the information to the individual.

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11. The method according to claim 9, wherein the scoring matrix comprises a combination of one or more scoring matrix vectors selected from the group consisting of a descriptor of family history, a descriptor of general medical physiological values, a descriptor of mRNA expression levels, a descriptor of methylation profiles, a descriptor of protein expression levels, a descriptor of enzyme activity, and a descriptor of antibody load.

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12. The method according to claim 9, wherein at least one of the one or more selection criteria is specified in advance by the individual.

13. The method according to claim 12, wherein at least one of the one or more selection criteria is a function of the availability of treatments effective to modify the phenotypic characteristic.

14. The method according to claim 9, wherein at least one of the one or more selection criteria is a function of the scope and quality of known research relating to the phenotypic characteristic.

15. The method according to claim 9, wherein at least one of the one or more selection criteria is a function of the probability determination(s) for one or more other phenotypic characteristics.

16. The method according to claim 9, further comprising, prior to communicating the information to the individual:

formatting the information relating to the relevant phenotypic attributes according to an organizational matrix, wherein the organizational matrix determines the grouping and presentation of information to the individual.

17. The method according to claim 16, wherein the organizational matrix groups phenotypic characteristics for which the individual has an enhanced probability together.

18. The method according to claim 16, wherein the organizational matrix groups phenotypic characteristics related to similar physiological systems together.

19. The method according to claim 16, wherein the organization matrix ranks the phenotypic characteristics as a function of the potential impact on the individual's lifestyle or quality of life.

20. The method according to claim 16, wherein the organization matrix ranks the phenotypic characteristics as a function of the genomic ethnicity of the individual.

21. The method according to claim 9, wherein prior to communicating the information to the individual, the identity of the individual is not associated with data corresponding to the genotypic characteristics, the relative probabilities of exhibiting the phenotypic characteristics, or the identified relevant information.

22. A method of evaluating the probability that progeny of two individuals of the opposite sex will exhibit one or more phenotypic attributes, the method comprising:

evaluating genomic markers from each of the two individuals for zygosity at each member of a preselected set of markers;

determining a probability distribution for the zygosity for each member of the preselected set of markers in the genomes of progeny of the two individuals;

comparing the probability distributions to a multivariate scoring matrix to obtain a probability distribution score, wherein the multivariate scoring matrix correlates patterns of marker zygosity with probabilities of exhibiting phenotypic attributes; and

determining whether the probability distribution score indicates that the progeny have an enhanced, diminished, or average probability of exhibiting one or more phenotypic attributes.

23. A method for determining the genomic ethnicity of an individual, comprising:

evaluating genomic markers from an individual at each member of a preselected set of markers;

comparing the genotype for each of the markers to a multivariate scoring matrix, wherein the multivariate scoring matrix correlates patterns of genotypes with probabilities of exhibiting phenotypic attributes; and

5 determining the genomic ethnicity of the individual as a pattern of the probabilities of exhibiting the phenotypic attributes.

24. The method according to any of claims 1, 2, 3, 4, 5, 6, 7 or 8, wherein the method is used for determining a genetic profile characteristic of a human population or subpopulation.

10 25. The method according to claim 24, wherein determining the genetic profile of the human population or subpopulation is used for pharmacogenomic analysis.

26. A method of selecting a set of genetic markers, comprising:

15 filtering one or more genetic markers for inclusion in the set by
determining measures of phenotypic value and/or prioritization selected from the group consisting of penetrance of the one or more markers in a population or subpopulation of interest, the degree of linkage of the one or more markers to a particular phenotype, the relative contribution of the one or more markers to communicating the phenotype, and the
20 degree of statistical or scientific confidence to be placed in any data associated with any of the measures of phenotypic value and/or priority used.